Information and Referral

Families of infants diagnosed with metabolic disorders may contact:

- Your health care provider
- South Dakota Department of Health Newborn Screening Program at 1-800-738-2301
- Children's Special Health Services Program at 1-800-738-2301
- The South Dakota Department of Health Newborn Screening Program homepage, with links to additional resources:

  http://www.state.sd.us/doh/NewbornScreening/index.htm

This pamphlet is available through the South Dakota Department of Health, Newborn Metabolic Screening Program, 615 E. Fourth St, Pierre, SD 57501-1700 or by calling (605) 773-3737

Or on the Department of Health website: http://www.state.sd.us/doh/NewbornScreening/index.htm

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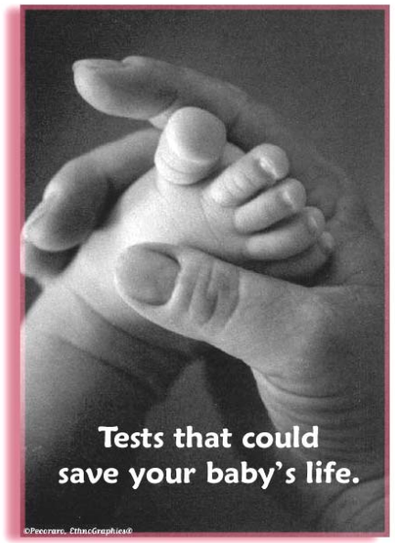
Newborn Screening



South Dakota Department of Health Newborn Screening Program

What is Newborn Metabolic Screening?

All newborns in South Dakota are required by law to have a blood test shortly after birth to screen for metabolic and other inherited disorders. The South Dakota Newborn Metabolic Screening Program helps identify babies who may have one of these disorders, and can alert the baby's doctor to the need for further testing and special care. With early diagnosis and medical treatment, complications from these serious but uncommon disorders can usually be prevented. This pamphlet was written to help answer your questions about the screening tests.

Who needs to be screened?

All infants born in South Dakota.

Why should my baby be screened?

Even if your baby looks healthy, he/she may have one of these disorders. Most infants born with metabolic disorders have no obvious signs at birth. Although these disorders are rare, they are usually serious. Without treatment babies with these disorders could suffer mental retardation, seizures, or even death.

How and when will my baby be screened?

A sample of blood is obtained by pricking the baby's heel. This sample is then placed on special paper which is sent to the designated laboratory for testing. The lab uses this one sample of blood to test for all the disorders. The sample is usually obtained on the day the baby is discharged from the hospital.

How will I learn of my baby's screening results?

The results of the tests are returned directly to your baby's doctor and the hospital where your baby was born. You can ask about the results when you take your baby in for a regular check-up. Generally, parents are notified only if retesting or further testing is needed. If your baby's doctor asks you to bring your baby in for retesting or further testing, do so as soon as possible.

What if my baby needs to be retested?

There are 3 main reasons why a repeat screening test may be needed: (1) there was a problem with the sample, (2) the test was done too early- prior to the 24 hours of age, or (3) the test result was abnormal. Generally, if the results of the repeat screening test are also abnormal, the doctor will discuss the need for further treatment or testing. It is important that your hospital and doctor have your correct address and phone number to contact you.

Optional Newborn Screening – Cystic Fibrosis

Cystic Fibrosis screening is available for infants born in South Dakota. The screening can be performed on specimens collected at the same time as the required newborn screening. Parents should visit with their doctor about ordering this test.

WHAT IS MY BABY SCREENED FOR?

DISORDER	WHAT IS IT?	WHAT HAPPENS IF NOT TREATED?	TREATMENT
Amino Acid and Acylcarnitine Screen ▪ **Amino Acid disorders (AA) disorders (includes Homocystinuria & Maple Syrup Urine Disease)**	These disorders are conditions that limit the body's ability to break down certain proteins.	Babies become very sick, become mentally retarded or may die.	Special diet.
▪ **Fatty acid oxidation (FAO) disorders (includes MCAD- medium chain acyl CoA dehydrogenase deficiency)**	These disorders affect the body's ability to make energy and to use the body's stored energy.	Can cause seizures, coma and even death.	Making sure infants and children are eating and drinking regularly, especially when they are ill.
▪ **Organic Acid (OA) disorders**	These disorders lead to high levels of acids in blood and urine.	Can cause seizures, mental retardation and death.	Special diet and medicine.
Biotinidase deficiency	The baby is unable to use the vitamin, biotin.	Babies grow very slowly and can become mentally retarded.	Daily biotin supplement.
Congenital Adrenal Hyperplasia	The body cannot make enough of certain types of hormones.	Affects growth and development. The body has trouble maintaining a balance of salt causing vomiting, dehydration, heart problems. If left untreated, death.	Special medicine.
Congenital hypothyroidism	The thyroid gland doesn't produce enough of the hormone thyroxine.	Babies do not grow and develop properly and become mentally retarded.	Medication – thyroxine.
Galactosemia	The body cannot use a certain sugar (galactose) found in dairy products.	Babies become very sick, have liver and eye damage, become mentally retarded or even die.	Special diet.
Hemoglobinopathies (Sickle Cell Anemia)	Red blood cells tend to change from the normal round shape to an abnormal sickle shape, which may cause blockage of blood flow.	Babies with sickle cell disease can get very sick and even die from common infections.	Antibiotic treatment may lessen problems.
Phenylketonuria (PKU)	The body cannot use a certain amino acid (protein) called phenylalanine.	Babies become mentally retarded.	Special diet.
Cystic Fibrosis (CF) **OPTIONAL TESTING**	Causes mucus to build up in the baby's lungs and intestines.	Causes problems with breathing and digesting food.	Regular medical care and a good diet.